ETFDH gene

electron transfer flavoprotein dehydrogenase

Normal Function

The *ETFDH* gene provides instructions for making an enzyme called electron transfer flavoprotein dehydrogenase. This enzyme is normally active in the mitochondria, the energy-producing centers in cells. Electron transfer flavoprotein dehydrogenase is involved in the process by which fats and proteins are broken down to produce energy.

Health Conditions Related to Genetic Changes

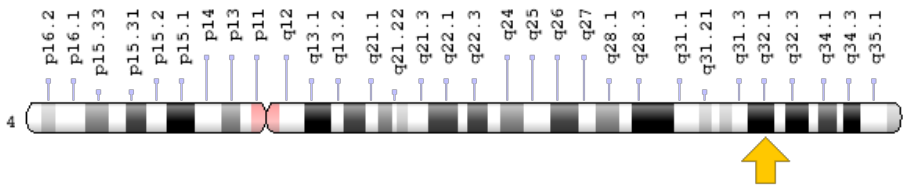
glutaric acidemia type II

Some mutations in the *ETFDH* gene prevent the production of the electron transfer flavoprotein dehydrogenase enzyme. Other mutations result in the production of a defective enzyme that cannot fulfill its role in the series of reactions (metabolic pathways) that break down fats and proteins. This enzyme deficiency allows these nutrients, as well as compounds created as the nutrients are partially broken down, to build up to abnormal levels, especially when the body is under stress. Toxic products of incomplete metabolism damage cells in many body systems, resulting in the signs and symptoms of glutaric acidemia type II.

Chromosomal Location

Cytogenetic Location: 4q32.1, which is the long (q) arm of chromosome 4 at position 32.1

Molecular Location: base pairs 158,672,101 to 158,708,713 on chromosome 4 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- electron transfer flavoprotein-Q oxidoreductases
- electron transfer flavoprotein ubiquinone oxidoreductase
- electron-transferring-flavoprotein dehydrogenase
- ETF dehydrogenase
- ETF-ubiquinone oxidoreductase
- ETFD_HUMAN
- ETFQO

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ETFDH%5BTIAB%5D%29+OR+%28electron-transferring-flavoprotein+dehydrogenase%5BTIAB%5D%29%29+OR+%28%28MADD%5BTIAB%5D%29+OR+%28ETFQO%5BTIAB%5D%29+OR+%28ETF-ubiquinone+oxidoreductase%5BTIAB%5D%29+OR+%28Electron+transfer+flavoprotein%5BTIAB%5D%29+OR+%28ubiquinone+oxidoreductase%5BTIAB%5D%29+OR+%28electron+transfer+flavoprotein+ubiquinone+oxidoreductase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2520+days%22%5Bdp%5D

OMIM

- ELECTRON TRANSFER FLAVOPROTEIN DEHYDROGENASE
  http://omim.org/entry/231675

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ETFDH%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=3483
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/2110
- UniProt
  http://www.uniprot.org/uniprot/Q16134

Sources for This Summary

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https://ghr.nlm.nih.gov/gene/ETFDH

Reviewed: July 2008
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
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